SHORT GCG EXPANSIONS IN THE PAB II GENE FOR OCULO-PHARYNGEAL MUSCULAR DYSTROPHY AND DIAGNOSTIC THEREOF

ABSTRACT OF THE DISCLOSURE

The present invention relates to a human PAB II gene containing transcribed polymorphic GCG repeat, which comprises a sequence as set forth in SEQ ID NO:3, which includes introns and flanking genomic sequence. The allelic variants of GCG repeat of the human PAB II gene are associated with a disease related with protein accumulation in nucleus, such as polyalanine accumula-tion, a disease related with swallowing difficulties, such as oculopharyngeal muscular dystrophy. The present invention also relates to a method for the diagno-sis of a disease with protein accumulation in nucleus, which comprises the steps of: a) obtaining a nucleic acid sample of said patient; and b) determining allelic variants of GCG repeat of the gene of claim 1, and wherein long allelic variants are indicative of a disease related with protein accumulation in nucleus.